

GENIE 8.0-public release notes

Sage Bionetworks

6/29/2020

Release Notes

- 16604 samples added
- New file formats
 - `data_guide.pdf`
 - `assay_information.txt`
- **NKI**
 - Updated all sample and patient ids. Sample and patient IDs from past releases are no longer valid and do not link/correlate to the current sample and patient IDs in the 8.2 release
 - Updated all RACE/ETHNICITY values from ‘Unknown’ to ‘Not Collected’.
- **YALE**
 - Intentionally reports only amplifications in copy number data due to internal policy.
 - **YALE-OCV-V2** panel’s expected gene count is 134 because one intron is included. Gene panels are created with only exons hence the difference in actual vs expected gene counts.
- **WAKE**
 - missing variants due to a Foundation Medicine conversion issue (Work in Progress)
- **UCSF**
 - Fixed genomic data issues.
- **CHOP**
 - All data removed due to known genomic artifacts.
- **PHS**
 - Fixed genomic data issues.
- **MSK**
 - Corrected oncotree code for some samples
- **COLU**
 - Fixed genomic data issues.
- Foundation Medicine genomic regions discrepancy
 - **DUKE/VICC/WAKE** use the Foundation Medicine T5A, T7, DX1, R2D2 bait sets, but some of the bed files uploaded don’t seem to match the expected gene count per panel.
- Sites/panels with *possible* genomic artifacts.
 - **CRUK-TS, JHU-500STP, UCHI-ONCOHEME55-V1, UHN-54-V1, VICC-01-MYELOID, WAKE, DUKE, COLU**
- Sites/panels with mismatching information for the number of genes in `assay_information.txt` vs `genomic_information.txt`.
 - **COLU, CRUK, DFCI, GRCC, JHU, MSK, NKI, PHS, SCI, UHN**
- Sites with missing panels in `assay_information.txt`
 - **UCHI**